TGFB2 gene

transforming growth factor beta 2

Normal Function

The *TGFB2* gene provides instructions for producing a protein called transforming growth factor beta-2 (TGFβ-2). This protein is found throughout the body and is required for development before birth and throughout life. To carry out its functions, TGFβ-2 attaches (binds) to receptor proteins on the surface of cells. This binding triggers the transmission of signals within cells, controlling various cellular activities. As part of a signaling pathway called the TGF-β pathway, the TGFβ-2 protein helps control the growth and division (proliferation) of cells, the process by which cells mature to carry out specific functions (differentiation), cell movement (motility), and controlled cell death (apoptosis). Because the TGFβ-2 protein keeps cells from growing and dividing too rapidly or in an uncontrolled way, it can suppress the formation of tumors.

The TGFβ-2 protein plays a role in the formation of blood vessels, the regulation of muscle tissue and body fat development, wound healing, and immune system function. TGFβ-2 is especially abundant in tissues that make up the skeleton, where it helps regulate bone growth, and in the intricate lattice that forms in the spaces between cells (the extracellular matrix).

Health Conditions Related to Genetic Changes

Loeys-Dietz syndrome

At least 20 mutations in the *TGFB2* gene have been found to cause Loeys-Dietz syndrome type IV. This disorder affects connective tissue, which gives structure and support to blood vessels, the skeleton, and many other parts of the body. Loeys-Dietz syndrome type IV is characterized by blood vessel abnormalities, heart defects, and skeletal deformities. The *TGFB2* gene mutations that cause this condition lead to the production of a TGFβ-2 protein with little or no function. As a result, the protein cannot bind to its receptors. Although the TGFβ-2 protein and its receptors are not bound, TGF-β pathway signaling occurs at an even greater intensity than normal. Researchers speculate that the activity of other proteins in this signaling pathway is increased to compensate for the reduction in TGFβ-2 activity; however, the exact mechanism responsible for the increase in signaling is unclear. The overactive

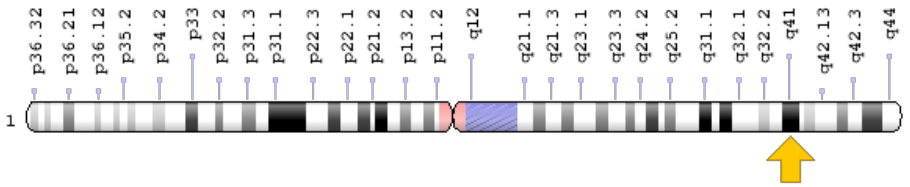
signaling pathway disrupts development of connective tissue and various body systems and leads to the signs and symptoms of Loeys-Dietz syndrome type IV.

A few mutations have been found that delete the entire *TGFB2* gene and some nearby genetic material. People with these deletions often have the features of Loeys-Dietz syndrome as well as features not usually associated with the condition, such as intellectual disability and movement problems. Researchers are working to determine which genes are missing as a result of these deletions and how their loss contributes to these additional signs and symptoms.

Chromosomal Location

Cytogenetic Location: 1q41, which is the long (q) arm of chromosome 1 at position 41

Molecular Location: base pairs 218,345,334 to 218,444,619 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- BSC-1 cell growth inhibitor
- cetermin
- G-TSF
- glioblastoma-derived T-cell suppressor factor
- polyergin
- TGF-beta2
- transforming growth factor beta-2
- transforming growth factor, beta 2

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Signal Proteins of the TGF-β Superfamily Act Through Receptor Serine/Threonine Kinases and Smads
  https://www.ncbi.nlm.nih.gov/books/NBK26822/#A2874
- Molecular Cell Biology (fourth edition, 2000): TGFβ signaling (image)
  https://www.ncbi.nlm.nih.gov/books/NBK21526/figure/A7151/

GeneReviews

- Loeys-Dietz Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1133

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TGFB2%5BTI%5D%29+OR+%28transforming+growth+factor+beta+2%5BTI%5D%29+OR+%28TGF-beta2%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- TRANSFORMING GROWTH FACTOR, BETA-2
  http://omim.org/entry/190220

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_TGFB2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=TGFB2%5Bgene%5D
- HGNC Gene Family: Endogenous ligands
  http://www.genenames.org/cgi-bin/genefamilies/set/542
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11768
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/7042
- UniProt
  http://www.uniprot.org/uniprot/P61812

Sources for This Summary

- Heldin CH, Landström M, Moustakas A. Mechanism of TGF-beta signaling to growth arrest, apoptosis, and epithelial-mesenchymal transition. Curr Opin Cell Biol. 2009 Apr;21(2):166-76. doi: 10.1016/j.ceb.2009.01.021. Epub 2009 Feb 23. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19237272
- Heldin CH, Moustakas A. Role of Smads in TGFβ signaling. Cell Tissue Res. 2012 Jan;347(1): 21-36. doi: 10.1007/s00441-011-1190-x. Epub 2011 Jun 4. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21643690
- Lindsay ME, Schepers D, Bolar NA, Doyle JJ, Gallo E, Fert-Bober J, Kempers MJ, Fishman EK, Chen Y, Myers L, Bjeda D, Oswald G, Elias AF, Levy HP, Anderlid BM, Yang MH, Bongers EM, Timmermans J, Braverman AC, Canham N, Mortier GR, Brunner HG, Byers PH, Van Eyk J, Van Laer L, Dietz HC, Loeys BL. Loss-of-function mutations in TGFB2 cause a syndromic presentation of thoracic aortic aneurysm. Nat Genet. 2012 Jul 8;44(8):922-7. doi: 10.1038/ng.2349.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22772368
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3616632/
- Ritelli M, Chiarelli N, Dordoni C, Quinzani S, Venturini M, Maroldi R, Calzavara-Pinton P, Colombi M. Further delineation of Loeys-Dietz syndrome type 4 in a family with mild vascular involvement and a TGFB2 splicing mutation. BMC Med Genet. 2014 Aug 28;15:91. doi: 10.1186/s12881-014-0091-8.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/25163805
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4236574/
- OMIM: TRANSFORMING GROWTH FACTOR, BETA-2
  http://omim.org/entry/190220

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/TGFB2

Reviewed: March 2017
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services